doctor think your baby may have one of these disorders. On rare occasions, your doctor may begin treatment before the results of the second test are known.

If you are asked to have your baby retested, please do so quickly! The earlier treatment is begun, the better the chance that a baby with one of these disorders will grow up healthy.

"Can my baby be cured if he or she has one of these disorders?"

At present, there is no "cure" for any of these disorders. However, the serious effects of these disorders can be lessened—and often completely prevented—if a special diet or other medical treatment is started early.

"How will I get my baby's test results?"

All test results are mailed to your doctor, and the hospital or other facility where your baby was born, within 14 days after the test was received at the laboratory. However, if any unusual results are found, your doctor is notified as soon as possible, usually by phone. For that reason, it is very important that you choose a doctor for your baby before he or she is born. Give the hospital the name of the doctor who will be taking care of your baby so that they can make sure your baby's doctor is listed on the newborn screening form.

Ask your doctor for your baby's test results at his or her first checkup. Along with newborn screening, well baby checkups are very important to make sure your baby is healthy and that problems are found before they become serious.

For more information, please contact:

South Carolina Department of Health
and Environmental Control
Women and Children's Services
Box 101106
Columbia, SC 29211
(803) 898-0767, or
your county health department.

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South Carolina Department of Health
and Environmental Control

ML #000032 PC (Rev. 5/00)

NEWBORN SCREENING

For Your Baby's Health

Parents sometimes worry about the health of their unborn baby. Many don't put those fears to rest until they are finally able to hold their newborn, count the fingers and toes, and see for themselves that their baby is perfectly healthy.

Usually, a newborn who *looks* healthy, *is* healthy. But sometimes, that may not be true. A baby may have "invisible" problems which could lead to mental retardation, abnormal growth, dangerous infections ... and even death.

Early detection of these "invisible" disorders is the goal of the South Carolina Department of Health and Environmental Control's Newborn Screening Program (South Carolina law requires this testing).

Through this program, all newborns are tested soon after birth for several genetic and chemical disorders. These disorders include *Phenylketonuria* (*called PKU*), *Congenital Hypothyroidism, Galactosemia, Congenital Adrenal Hyperplasia (called CAH*), *Medium Chain Acyl Co-A Dehydrogenase Deficiency (called MCADD),* and *Hemoglobinopathies.* The tests are done on a small sample of blood taken by pricking the baby's heel. Early treatment can give an infant with one of these disorders the best chance for a healthy, productive life.

"What is each of these disorders?"

PKU is a disorder that keeps the baby's body from being able to use certain parts of the proteins (amino acids) found in milk and formula. This amino acid (phenylalanine) builds up in the baby's system and can damage growing brain cells, causing mental retardation. Doctors can give babies with PKU a special formula and diet low in phenylalanine.

Congenital Hypothyroidism means that the thyroid gland is not working properly. If untreated, a baby with Congenital Hypothyroidism will not grow or function normally and may develop severe mental retardation.

Galactosemia is a condition where the baby cannot use a sugar (galactose) found in cow's milk-based infant formula and breast milk. Babies who are not treated can develop life-threatening infections and mental retardation. This disorder can be treated by feeding the baby soy-based infant formula.

In babies with **CAH**, the body's adrenal gland does not work normally. Untreated babies will not grow or mature properly. Some of these babies may even die.

The body uses a sugar called glucose as the main energy source. When the glucose cannot be used, fat is broken down for energy. **MCADD** is a disorder where the body is unable to use certain kinds of fat to make energy. Because they cannot use fat for energy, babies with **MCADD** may get very sick if they have an illness that makes them not want to eat. They can have trouble breathing and have seizures. Their hearts may even stop beating if their blood sugar gets too low. The main treatment for MCADD is to make sure the baby eats every few hours.

Hemoglobinopathies are genetically caused hemoglobin disorders such as Sickle Cell Anemia. These disorders can cause many problems including misshaped red blood cells, anemia, severe pain and high risk for serious infection.

"What if my new baby seems very healthy? Are these tests really necessary?"

The tests are necessary. Most babies who have PKU, Congenital Hypothyroidism, Galactosemia, CAH, MCADD or Hemoglobinopathies seem healthy at birth. Most are born into families who have no history of genetic or chemical disorders. Blood tests are the only way these disorders can be found in the early stages.

"These disorders seem rare. Why is there a state law requiring screening?"

PKU, Congenital Hypothyroidism, Galactosemia, CAH, MCADD and Hemoglobinopathies are uncommon, but they are also very serious. Testing every baby at birth is the best way to make sure all babies who have these disorders are found quickly and treated as soon as possible.

"What does it mean if I'm told my baby needs a second test?"

Retesting may be needed for a number of reasons. Sometimes a retest is needed simply because enough blood was not collected the first time. Babies whose first blood sample is taken before they are 24 hours old should also have a second test as a precaution.

Also, the tests are very sensitive to make sure a baby who really has one of these disorders will not be missed. Because of this, a few normal babies will have false positive results and will need a second test. While taking your baby in for repeat testing can be scary, it is important that every baby has a thorough screening. As a general rule, only when a baby's test is unusual for a second time will your